ALMS1 gene

ALMS1, centrosome and basal body associated protein

Normal Function

The *ALMS1* gene provides instructions for making a protein whose function is unknown. Researchers believe that the protein may play a role in hearing, vision, regulation of body weight, and functions of the heart, kidney, lungs, and liver. It may also affect how the pancreas regulates insulin, a hormone that helps control blood sugar levels.

The ALMS1 protein is present in most of the body's tissues, usually at low levels. Within cells, this protein is located in structures called centrosomes. Centrosomes play a role in cell division and the assembly of microtubules, which are proteins that transport materials in cells and help the cell maintain its shape. The ALMS1 protein is also found at the base of cilia, which are finger-like projections that stick out from the surface of cells. Almost all cells have cilia at some stage of their life cycle. Cilia are involved in cell movement and many different chemical signaling pathways. Based on its location within cells, researchers suggest that the ALMS1 protein might be involved in the organization of microtubules, the transport of various materials, and the normal function of cilia.

Health Conditions Related to Genetic Changes

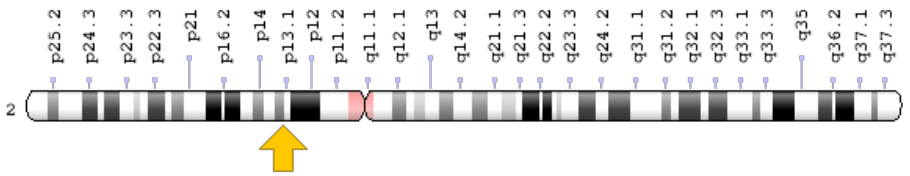
Alström syndrome

More than 80 mutations in the *ALMS1* gene have been identified in people with Alström syndrome. Most of these mutations lead to the production of an abnormally small version of the ALMS1 protein that does not function properly. Researchers propose that a lack of normally functioning ALMS1 protein in the brain could lead to overeating. A loss of this protein in the pancreas may cause insulin resistance, a condition in which the body cannot use insulin properly. The combined effects of overeating and insulin resistance impair the body's ability to handle excess sugar, leading to diabetes and obesity (two common features of Alström syndrome). It is unclear how *ALMS1* mutations cause the other signs and symptoms of Alström syndrome. Researchers suspect that this condition is associated with malfunctioning cilia in many of the body's tissues and organs.

Chromosomal Location

Cytogenetic Location: 2p13.1, which is the short (p) arm of chromosome 2 at position 13.1

Molecular Location: base pairs 73,385,758 to 73,609,919 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ALMS1_HUMAN
- Alstrom syndrome 1
- Alstrom syndrome protein 1
- KIAA0328

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Cilia and Flagella Are Motile Structures Built from Microtubules and Dyneins
  https://www.ncbi.nlm.nih.gov/books/NBK26888/#A3075
- The Cell: A Molecular Approach (second edition, 2000): Structure, Assembly, and Dynamic Instability of Microtubules
  https://www.ncbi.nlm.nih.gov/books/NBK9932/#A1821

GeneReviews

- Alstrom Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1267

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALMS1%5BTIAB%5D%29+OR+%28KIAA0328%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- ALMS1 GENE
  http://omim.org/entry/606844

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALMS1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALMS1%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=428
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7840
- UniProt
  http://www.uniprot.org/uniprot/Q8TCU4

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https://ghr.nlm.nih.gov/gene/ALMS1

Reviewed: August 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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